

REVIEW

Emotional experience of the diagnostic process of a rare disease and the perception of support systems: A scoping review

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Abstract

Aims and objective: To explore the experience of adult patients and adult patients' families, and their perception of the support systems received during the diagnostic process of rare diseases.

Background: There are about 7,000 rare diseases that affect 7% of the world's population. Rare diseases are often underdiagnosed. This has been reported to have deleterious physical and psychological consequences in both the patients and their families, especially when institutional support during this process is low.

Design: A scoping review was carried out following the 6-phase model proposed by Arksey & O'Malley and Levac *et al.*, including the consultation phase in which patients diagnosed with rare diseases were interviewed to seek their views on the bibliographic evidence reviewed and their experience during the diagnostic process.

Methods: The databases consulted were PubMed, CINAHL, Web of Science, SCOPUS, Cochrane Library, PsycINFO, OpenGrey, ProQuest Dissertations and Theses Global. They were explored from inception–July 2020, and qualitative, quantitative and mixed method studies were included. The Mixed Methods Appraisal Tool was used for the critical evaluation of the articles. The review was based on the guidance in the PRISMA-ScR statement.

Results: The initial search identified 2,350 articles, of which 20 fully met the inclusion criteria and were therefore reviewed. In this analysis appeared two dimensions: internal factors: emotional aspects, and external factors: resources and support systems.

Relevance to clinical: This review provides evidence on the emotional impact of the diagnostic process and during the communication phase of the definitive diagnosis. Health systems and professionals must be strengthened in order to improve the information, training and resources. Nurses can play a key role in coordinating communication and follow-up of those affected.

KEYWORDS

diagnostic, emotional, experiences, rare disease, support system

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1 | INTRODUCTION

Rare diseases (RDs) are defined as those that have a low prevalence in the population. To qualify as rare, each specific disease can only affect less than 5 in 10,000 inhabitants (Aymé et al., 2008; Bogart & Irvin, 2017) such as myelofibrosis with a prevalence of 2,7: 100.000 individuals (Alarcón-Ovalle et al., 2016) or primary ciliary dyskinesia whose reported prevalence varies from 1: 2000–1: 40000 (Behan et al., 2016) and Hermansky–Pudlak syndrome with a prevalence of 1: 500.000 a 1: 1.000.000 in non-Hispanic people and 1: 18.00 in Hispanic people (Christensen et al., 2017).

Despite the differences in the aetiology and symptoms of RDs, many of them are chronic, involve multisystemic dysfunction, are not effectively treated and require complex care (Aymé et al., 2008). Due to the small amount of people affected by each individual RD, funding for research into causes and treatments is limited, which slows down the discovery of diagnostic tools and potential therapies (Grigull et al., 2019).

There are also psychological consequences, the name itself indicates it, "rare disease" and that is why many people who suffer from it feel out of the ordinary (Blöß et al., 2017). As a result, they feel discriminated at a social level in aspects such as leisure, education or even daily life activities (Alarcón-Ovalle et al., 2016). Affected people need help with daily activities and often lack the resources to address these needs (Budych et al., 2012).

The diagnosis of an RD helps facilitate access to effective medical, psychological and social care and treatment (Aymé et al., 2008). Getting the right diagnosis, even when there is no treatment, increases people's opportunities to plan for their future (Khosla & Valdez, 2018). The moment of diagnosis is a painful experience in most cases (Bogart & Irvin, 2017). Being diagnosed with an RD is a long road, and the urgency of the patient and their family members does not always coincide with the speed observed by professionals or the health system (Heuyer et al., 2017). The process is called the diagnostic journey which starts when patients first go to a health-care provider to consult their symptoms and ends when they receive the correct diagnosis. This journey is fraught with great emotional and physical baggage (Geng et al., 2019).

In this process, the lack of information about RDs, the lack of co-ordination between health professionals, the lack of specialists with clinical experience and the poor functioning of the health services affect both the person and his/her family negatively from the first moment (Crowe et al., 2019). Jaeger et al., (2015) showed in their study that people with RDs are isolated and most of the population lacks awareness about them, resulting in their being misunderstood, avoided and blamed. Consequently, social and environmental barriers limit the ability to participate in social roles and activities (Bogart & Irvin, 2017). Similarly, the psychological and emotional reactions associated with the diagnosis can negatively influence the process of adaptation to the disease and the quality of life (Castillo-Esparcia & López-Villafranca, 2016). In reality, these emotions are the adjustment to a multi-component process that generates a response to the adaptive challenges of a particular situation (Nesse & Ellsworth, 2009).

What does this paper contribute to the wider?

- There is a deficit of care for those affected, as opposed to chronic diseases that are not rare; therefore, there is a need for multidisciplinary support; but in the case of RD, the professionals who would have to provide this support also have relevant deficiencies that must be addressed.
- The emotional aspects that have emerged require the development of basic strategies such as patient-professional communication and trust, as well as large-scale research and early diagnosis strategies, especially those that minimise the impact of socio-demographic and gender aspects that act as health determinants in RD.
- Interventions and studies in the field of RD nursing are essential, as is the need to strengthen the professional role in the training and care of individuals and families affected by RD.

This study has focused on the diagnostic process, that is on the two initial phases described by Schwartz (2009), which both the person and his/her family environment go through. There are 5 phases: (a) uncertainty and confusion due to the appearance of the first symptoms; (b) confusion at the time the diagnosis is received; (c) opposition and isolation, as well as cushioning to maintain hope and to be able to fit the new reality; (d) anger and sadness; and finally (v) phase of adaptation. For the people affected and their families, going through these phases has emotional implications generated by stressful life circumstances. Additionally, due to the low prevalence of the different RDs studied individually there is a lack of knowledge and holistic perspective of service providers (Budych et al., 2012). These systems are classified as inadequate to respond to the needs of patients, and this inadequacy leads to unpleasant experiences.

Finally, it should be noted that there is a lack of literature reviews that explore the psychological theme. After reviewing the literature, we found the study by von der Lippe et al., (2017) which addresses the psychosocial experience of adults who share a diagnosis of a rare disease through a systematic review. However, there are no reviews that focus exclusively on the pre-diagnosis phase and the time of diagnosis. Knowing the emotional experience (affected persons and family members) in these two phases through the detailed overview provided by a scoping review can undoubtedly help to achieve a better adaptation of those affected in these and subsequent stages.

2 | AIMS

This scoping review aims to explore, according to published evidence, the emotional experience of adults with an RD and their families during the diagnostic process and their perception of the

support activities received to meet their health, care or daily life needs.

3 | METHODS

3.1 | Study design

This scoping review used the Arksey and O'Malley (2005) framework. Subsequently, other authors (Levac et al., 2010) proposed supplementary considerations to the review model, adding a more complete explanation of each of the six stages of the process. The scoping review followed the Preferred Reporting Items for Systematic Review and Meta-Analyses extension for Scoping Review (PRISMA-ScR) checklist (File S1).

3.2 | Stage 1: identifying the research question

Based on Levac et al. (2010), research questions should consider justification and clarify purposes. To this end, it was agreed that the following questions should be asked:

- How do adults with an RD and/or their families describe their experiences during the diagnostic process?
- What is the perception of adults with an RD and/or their families about the support activities they were provided with to meet their care needs during the diagnostic process?

3.3 | Stage 2: identifying relevant studies

The databases consulted for the identification of studies were PubMed, CINAHL, Web of Science, SCOPUS, Cochrane Library and PsycINFO, and for grey literature, Google, Google Scholar, OpenGrey and ProQuest Dissertations and Theses Global. A manual review of article references was conducted as an additional data source.

A comprehensive search strategy was developed by an information specialist (see File S2 for search strategy). The appropriate keywords were selected for each database, using MeSH or free terms: Diagnosis, Experiences, Rare Disease and Support System, among others, as well as Boolean operators for maximum sensitivity and specificity.

Eligibility criteria included the following: (a) studies of any type of RD (qualitative, quantitative and mixed method studies) were systematically searched from inception–31 July 2020; (b) study population: adults (16+) with an RD diagnosis, family members and experts (healthcare professionals); (c) articles related to the experience of the disease from the patient and/or family perspective; (iv) articles focusing on psychosocial and support care, access to services, quality of care, experiences, daily activities and consequences of delayed diagnosis; and (v) publications in English, Spanish, Italian, French and Portuguese. Exclusion criteria included the following: (a) articles

funded by commercial interests and (b) review studies of the literature, letters, conference papers or similar primary documents.

3.4 | Stage 3: study selection

The selection process was carried out using the Covidence tool (<https://www.covidence.org/>). This tool is web-based software used to do primary screening, data extraction and synthesise. This process was carried out by two independent reviewers (LL and AT). Any differences were resolved by a third reviewer (JR). This third reviewer examined 10% of the abstracts for eligibility criteria.

Following the guidelines of Levac et al. (2010), the rest of the research team met at the initial, intermediate and final stages of the review process to resolve any uncertainties related to the selection of studies. Though is not mandatory in a scoping review (Arksey & O'Malley, 2005), a decision was made to assess methodological quality using the Mixed Methods Appraisal Tool (MMAT)—2018 Version. The choice of this tool was based on the fact that it includes all types of methodological studies (quantitative, qualitative and mixed).

3.5 | Stage 4: charting the data

A standardised, pre-piloted form was used to extract data from the included studies. The following information was extracted: (a) author; (b) year of publication; (c) language; (iv) country of study; (v) characteristics of the population studied; (vi) quality of the study; (vii) purpose of the study; and (viii) main results.

One review author extracted the data (LL), and this was cross-checked by another member of the review team.

3.6 | Stage 5: collating, summarising and reporting the results

The included manuscripts were manually coded into themes, which were grouped into the following dimensions: (a) internal factors: emotional aspects and (b) external factors: resources and support systems.

3.7 | Stage 6: consultation

This consultation stage is optional. Interviews with patients with RD were organised through the Spanish Federation of Rare Disease that included the profiles of adults with a diagnosis no older than 10 years. This period was considered for the proximity to the diagnostic event, and to ensure the confirmability of the information. The interviews addressed the pre-diagnosis experience and the experience at the time of diagnosis based on aspects such as (a) personal, family and social experience; and (b) resources and support

used or available. This phase was used to validate the results of the review (Arksey & O'Malley, 2005; Levac et al., 2010).

3.8 | Ethical aspects

Given the incorporation of phase 6 in the review, the Clinical Research Ethics Committee of the Hospital Universitari Arnau de Vilanova de Lleida (Spain) was consulted. The issued resolution stated that, given the nature of the study, it did not require their approval.

The participants in the consultation phase signed the informed consent to participate in the project. Data confidentiality and anonymity was ensured throughout the process by assigning an alpha-numeric code to each interview. All participants had the opportunity to review the content of their interviews and make comments.

4 | RESULTS

4.1 | Study selection

The initial search identified a total of 2,350 articles. Applying the inclusion and exclusion criteria, indicated in stage 2 of the scoping review methodology, the number of studies was reduced to 1,214. Subsequently, screening of titles/abstracts was used and 161 remained for full text selection. Specifically, of the

141 studies, 88 were found to be inappropriate, 46 were on the wrong subject and in 7 the language was not in the inclusion criteria. Finally, 20 articles were considered eligible for inclusion in the data extraction.

Likewise, in the concordance analysis between evaluators, 97% (p1) of coincidences were observed as opposed to the 85% (pe) expected by chance, in this case $p1 > pe$, so we found more concordance than was expected by chance, placing it at a Cohen kappa index of 0.810. This evaluation was almost perfect.

The flow of literature through the study is shown in Figure 1.

4.2 | Description of included studies

The results in relation to the description of the studies are organised in two points: characteristics of the included studies and quality of the evidence of the studies. The main characteristics of the studies are shown in File S3.

4.2.1 | Study characteristics

The studies were carried out in countries on different continents, especially in Europe (Germany, Denmark, Spain, Italy, Portugal and the UK) 50% ($n = 10$), followed by America (Brazil, Canada, Colombia and the US) 35% ($n = 7$), then Oceania (Australia) 10% ($n = 2$) and finally Asia (China) with 5% ($n = 1$).

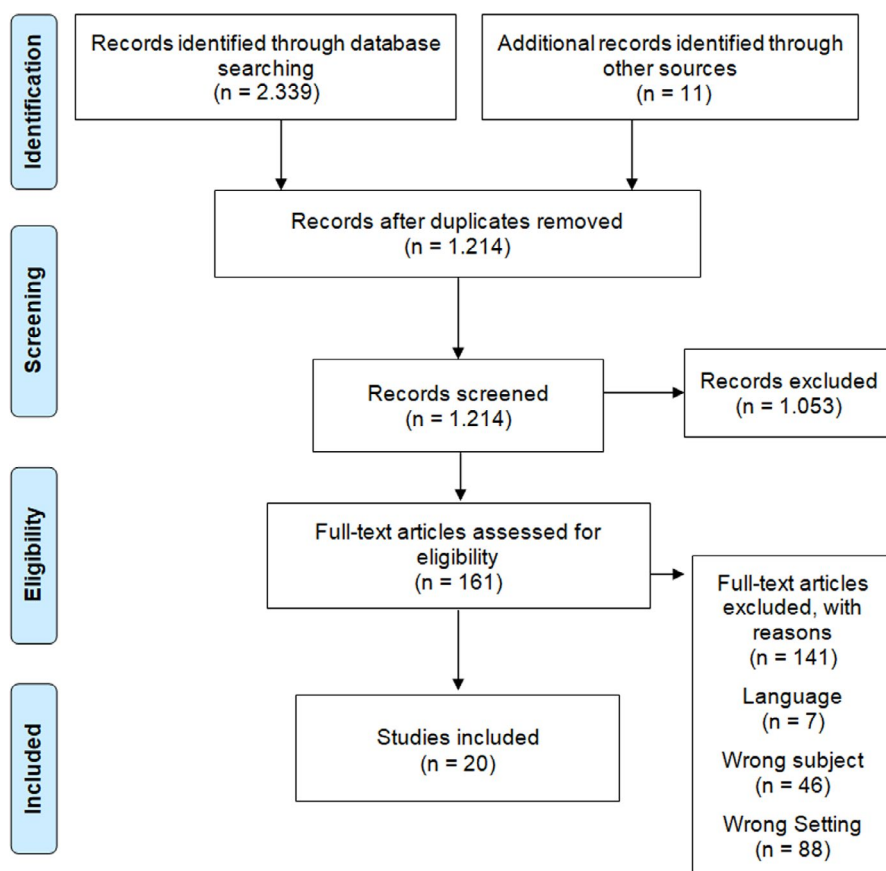


FIGURE 1 PRISMA diagram

The subjects included in the research had different roles in relation to the RD, that is patients, relatives and/or professionals. Of the twenty studies, four of them, or 20%, showed evidence focused only on patients diagnosed with an RD (Adams et al., 2018; Alarcón-Ovalle et al., 2016; Christensen et al., 2017; Jensen et al., 2019), and 45%, nine studies of the total, reported only on relatives (Anderson et al., 2013; Bendixen & Houtrow, 2017; Cardinali et al., 2019; Custódio et al., 2018; Esteban et al., 2015; Germini et al., 2018; Granero-Molina et al., 2020; Lim et al., 2012; Zurynski et al., 2017). Then, four studies focused on both family members and patients with an RD, representing 20% (Behan et al., 2016; Esquivel-Sada & Nguyen, 2018; Henderson et al., 2009; Requena et al., 2014), whereas only one study focused on RD experts (Blöß et al., 2017). One included patients, relatives and lawyers (Kesselheim et al., 2015), and one included patients, relatives, organisations and healthcare professionals related to RDs which represent 5% each (Lopes et al., 2018). From this description, we can see that 75% of the articles studied the relatives, 50% included the affected people, and only 10% considered the professionals or associations that offer care to people with an RD.

The total number of participants was 1568, men (81.7%) and women ($n = 18.3\%$). The age of the participants with an RD was 16 years minimum and 80 years maximum.

Some of the studies (Blöß et al., 2017; Cardinali et al., 2019; Esquivel-Sada & Nguyen, 2018; Kesselheim et al., 2015; Lopes et al., 2018; Requena et al., 2014; Zurynski et al., 2017) refer to RD pathologies in general, although others detail the pathologies such as lipodystrophy (Adams et al., 2018), myelofibrosis (Alarcón-Ovalle et al., 2016), lysosomal storage disease and mitochondrial disease (Anderson et al., 2013), primary ciliary dyskinesia (Behan et al., 2016), duchenne muscular dystrophy (Bendixen & Houtrow, 2017), Hermansky-Pudlak syndrome (Christensen et al., 2017), Wolfram's syndrome (Esteban et al., 2015), Bartter syndrome (Germini et al., 2018), hereditary angioedema (Granero-Molina et al., 2020), Niemann-Pick disease type B (Henderson et al., 2009), mastocytosis (Jensen et al., 2019), Rett syndrome (Lim et al., 2012) and osteogenesis imperfecta type 1 (Custódio et al., 2018).

The level of education is only mentioned in 6 studies (Cardinali et al., 2019; Christensen et al., 2017; Esquivel-Sada & Nguyen, 2018; Germini et al., 2018; Henderson et al., 2009; Lim et al., 2012). Three studies (Alarcón-Ovalle et al., 2016; Cardinali et al., 2019; Henderson et al., 2009) included assessment of the socio-economic conditions of the participants, detailing the different economic strata (high, medium and low) and total yearly income.

The ethnicity of the participants is only collected in the study by Anderson et al. (2013). Likewise, the articles do not refer to the cultural aspects of the participants. In the study by Henderson et al. (2009), they do value religion.

4.2.2 | Methodological quality

The review comprises 20 studies of qualitative methodology (75%), quantitative methodology (10%) and mixed methods (15%). All were

critically evaluated according to the MMAT tool. These judgements are presented in File S4. In the following section, methodological quality is summarised narratively based on the study design.

Qualitative studies ($n = 14$) (Lim et al., 2012; Requena et al., 2014; Kesselheim et al., 2015; Alarcón-Ovalle et al., 2016; Bendixen & Houtrow, 2017; Blöß et al., 2017; Christensen et al., 2017; Adams et al., 2018; Lopes et al., 2018; Esquivel-Sada & Nguyen, 2018; Germini et al., 2018; Cardinali et al., 2019; Jensen et al., 2019; Granero-Molina et al., 2020). All studies were judged to have appropriately used the qualitative approach to answer the research question and to have adequately used the qualitative data collection methods to address the research question. One study (Requena et al., 2014) does not specify the data analysis used. The interpretation of results of one study (Alarcón-Ovalle et al., 2016) is not supported by the data collected. All studies show clear links between data sources, collection, analysis and interpretation.

Quantitative descriptive studies ($n = 2$) (Anderson et al., 2013; Zurynski et al., 2017). These studies were judged to be employing a relevant sampling strategy and appeared to have used appropriate measurements. However, the study (Anderson et al., 2013) judged that the sample was not regarded to be representative of the target population. One study (Zurynski et al., 2017) was judged as low risk of nonresponse bias. Both studies show clearly established and justified statistical analysis.

Mixed methods studies ($n = 4$) (Behan et al., 2016; Custódio et al., 2018; Esteban et al., 2015; Henderson et al., 2009). All studies show adequate justification for using a mixed method design and effectively integrate study components to address the research question. The four studies do not show divergences or inconsistencies when integrating the findings of the qualitative and quantitative components. However, one study (Esteban et al., 2015) does not explain how the data were extracted from the qualitative component.

4.3 | Experiences and perceptions of patients and families

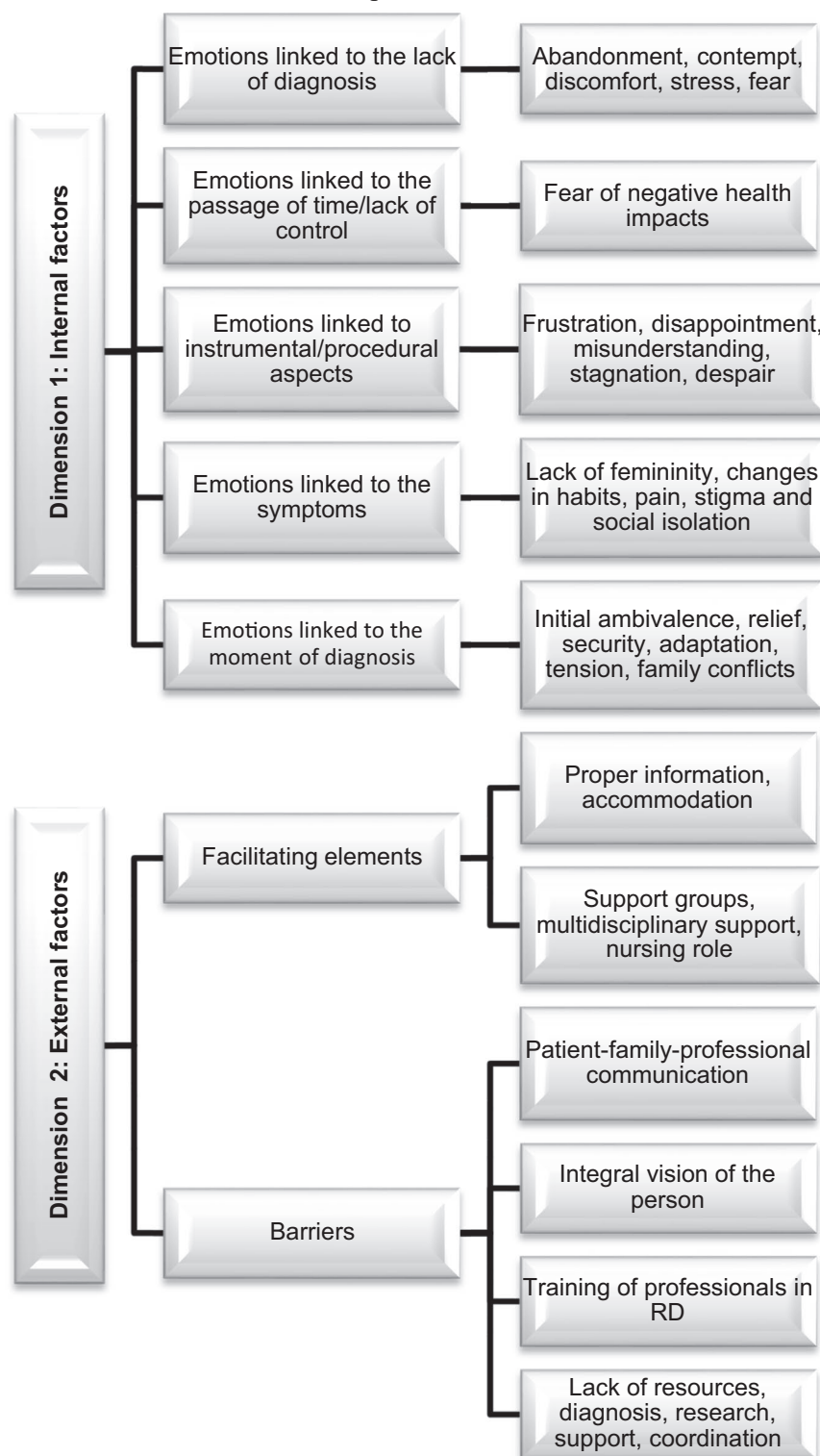
This review showed findings about the diagnostic care process experienced and perceived by people affected by RD and their families. The dimensions explored were internal factors (personal aspects of the experience and impact of the diagnosis) and external factors (resources and support systems). In this sense, a series of themes emerged which allowed us to detail the origin and nature of the emotional reactions throughout the diagnostic process. In Figure 2, a scheme of findings is shown.

4.3.1 | Internal factors: Emotional aspects

Emotions linked to the lack of diagnosis

Emotions linked to the lack of diagnosis were sometimes shown as feelings of abandonment or contempt, when they were not taken into account or taken seriously (Behan et al., 2016; Esquivel-Sada

FIGURE 2 Matrix of findings



& Nguyen, 2018; Kesselheim et al., 2015); experiences doubly reported by patients and their families (Bendixen & Houtrow, 2017; Zurynski et al., 2017). They related this shared process (person-environment) and this multidimensional experience of psychological, social and physical discomfort.

There was even a relationship between stress and information: the more stress, the more pressing the need to know (Germeni

et al., 2018). Families also reported feelings of guilt about the lack of a diagnosis (Granero-Molina et al., 2020) or the fear of not feeling competent to care for their child (Custódio et al., 2018).

Emotions linked to the passage of time and lack of control

Emotions linked to the passage of time and lack of control were associated with undiagnosed disease progression and fear of

negative health impacts due to delay in diagnosis (Behan et al., 2016; Christensen et al., 2017; Granero-Molina et al., 2020; Kesselheim et al., 2015; Zurynski et al., 2017).

Emotions linked to instrumental or procedural aspects

Emotions linked to instrumental or procedural aspects were linked to the need to live through years of examinations and difficulties, transfers, hospitalisations, surgery or various treatments (Alarcón-Ovalle et al., 2016; Germeni et al., 2018; Granero-Molina et al., 2020; Lim et al., 2012), as well as receiving misdiagnosis and unnecessary treatment (Behan et al., 2016; Kesselheim et al., 2015). As described above, the study by Blöß et al. (2017) detailed that RD patients shared pre-diagnostic phenomena that included a high degree of frustration. That is, common experiences that included a feeling of disappointment and being incorrectly labelled as having psychological problems or being hypochondriacal (Jensen et al., 2019). Requena et al., (2014) described it as a negative circular process of stagnation and pilgrimage in the search for a diagnosis. Sometimes, alternative methods were used without any scientific basis because of desperation in the search for answers (Germeni et al., 2018).

The lack of therapeutic communication in the absence of a diagnosis was an exhausting process, on a psychological, social and even health professional level, which lead to a great deal of misunderstanding for the individual and his/her family (Granero-Molina et al., 2020). Sometimes, the doctors did not accept the complaints and/or these were misinterpreted (Blöß et al., 2017) and even confrontations occurred (Requena et al., 2014).

Emotions linked to the symptomatology

Emotions linked to the symptomatology according to the pathology. These signs and symptoms were diverse due to the large number of diseases and their characteristics. The disease was unpredictable and difficult to control (Jensen et al., 2019). As an example, in the study by Custódio et al. (2018) the threat of pain from fractures was continuous or in the study by Adams et al. (2018) on lipodystrophy due to physical changes, female participants described an impact on their femininity; they felt inferior because they did not meet the expectations of how a woman should be. This discomfort with their bodies leads to negative feelings about sexuality (Jensen et al., 2019).

They also referred to changes in aspects of daily life such as eating, sleeping, working and physical activity (Alarcón-Ovalle et al., 2016; Henderson et al., 2009; Lopes et al., 2018). Sometimes, suffering from RD leads to isolation and social stigma due to rejection (Granero-Molina et al., 2020; Henderson et al., 2009; Lopes et al., 2018), a situation that was exacerbated in women (Jensen et al., 2019). These derived emotional aspects such as frustration and self-doubt went completely unnoticed by health professionals (Blöß et al., 2017) as they focused more on physical symptoms.

Emotions linked to the moment of diagnosis appear in studies

Emotions linked to the moment of diagnosis appear in studies (Adams et al., 2018; Behan et al., 2016; Christensen et al., 2017; Germeni

et al., 2018; Granero-Molina et al., 2020; Jensen et al., 2019; Lopes et al., 2018) where they related the relief of diagnosis as a source of explanation for their medical problems, but also of the generation of negative feelings such as anxiety, stress, sadness, anger, fear, guilt, disbelief, denial, avoidance or concealment (Alarcón-Ovalle et al., 2016; Esquivel-Sada & Nguyen, 2018; Esteban et al., 2015; Zurynski et al., 2017). Thus, the impact of the diagnosis was multiple since it influenced at a personal and social level (Esquivel-Sada & Nguyen, 2018). Patients highlighted the ambivalent feelings at the beginning, and later, the relief.

This process was shared in other studies (Adams et al., 2018; Germeni et al., 2018; Granero-Molina et al., 2020; Jensen et al., 2019; Lopes et al., 2018), which referred to being diagnosed as creating trust, meaning, order, adaptation and security in a chaotic number of symptoms for patients and families, in addition to the security of being able to follow adequate treatment and gaining access to different specialists (Esquivel-Sada & Nguyen, 2018) and to stop feeling misunderstood, intimidated and stigmatised (Granero-Molina et al., 2020).

The people affected and their families remembered the moment of the diagnosis in detail; the space, the verbal and non-verbal communication, and the emotions experienced. Without a doubt, the moment of diagnosis was a time of tension for everyone involved as an RD implies a infrequent and chronic disease which can sometimes be degenerative and without available treatment (Requena et al., 2014).

It is also important to note that a diagnosis of a genetic disease could lead to family conflicts (such as the difficulty of sharing the diagnosis with other members for fear of repercussions or discrimination because of an affected family lineage), and at the same time, it was shown to be an aid to life planning and reproductive decision-making (Esquivel-Sada & Nguyen, 2018; Esteban et al., 2015).

4.3.2 | External factors: Resources and support systems

Two themes emerged from the external factors: facilitating elements and barriers.

Facilitators

Facilitators, according to the study by Granero-Molina et al. (2020), were channelled through the establishment of support groups and the provision of appropriate information; these maximised emotional, social and instrumental support. Patients confirmed support from family, support groups, specialised associations (Anderson et al., 2013; Germeni et al., 2018; Zurynski et al., 2017) and spiritual support groups (Alarcón-Ovalle et al., 2016; Christensen et al., 2017). They also highlighted the benefits of sharing experiences with peer groups, in social networks or in face-to-face meetings (Adams et al., 2018; Christensen et al., 2017; Esquivel-Sada & Nguyen, 2018).

Other elements such as the need for financial support and health services with a multidisciplinary character (doctors, nurses,

psychologists, social workers and/or case managers) (Anderson et al., 2013; Zurynski et al., 2017) were also mentioned. In the study by Granero-Molina et al. (2020), the role of community nurses was highlighted. They were able to help to improve knowledge and train patients and families in drug administration and different care needs.

At the time of diagnosis, it was essential to balance the information being supplied (Requena et al., 2014). Families shared this aspect, and therefore, the need for adequate information and training enabled them to provide care (Custódio et al., 2018). This helped a two-tier accommodation process to take place: from the point of view of content and of the emotions generated (Requena et al., 2014).

Finally, and a very relevant facilitator to be promoted, was the collaborative and proactive attitude of people with an RD (Requena et al., 2014).

Barriers

Barriers were defined as those aspects that hindered the process or needed to be improved given their relevance to the diagnostic process. A very important element was the need to improve communication between patient and health professional (Requena et al., 2014; Bendixen & Houtrow, 2017; Custódio et al., 2018; Esquivel-Sada & Nguyen, 2018; Cardinali et al., 2019). On the other hand, the people affected put forward the need for a more global vision among specialists.

This aspect of the need for multidisciplinary and coordinated comprehensive care from the outset was reported by other studies (Bendixen & Houtrow, 2017; Lopes et al., 2018; Zurynski et al., 2017). Lack of guidance during diagnosis broke the trust between healthcare personnel, patients and families (Bendixen & Houtrow, 2017; Cardinali et al., 2019).

As a key element, the limited knowledge of doctors (Adams et al., 2018; Anderson et al., 2013; Cardinali et al., 2019; Henderson et al., 2009; Kesselheim et al., 2015; Lim et al., 2012; Lopes et al., 2018; Zurynski et al., 2017) and nurses (Lopes et al., 2018) should have been noted. Therefore, it was essential to promote training in RDs at the undergraduate, postgraduate and continuing education levels (Anderson et al., 2013). This lack of knowledge and training meant that existing evidence-based diagnostic standards and clinical guidelines for some pathologies were not applied (Bendixen & Houtrow, 2017). Even in the study by Christensen et al. (2017), participants narrated their role as experts and the need they felt to educate professionals, in a role reversal. This element was also made explicit in other studies (Kesselheim et al., 2015).

Other barriers included inadequate and insufficient resources (Germeni et al., 2018; Granero-Molina et al., 2020; Henderson et al., 2009), the presence of experts in tertiary centres and not in the community (Kesselheim et al., 2015) which forced long waits and very long journeys (Lim et al., 2012), the difficulty of diagnosing diseases not included in screening and the lack of warning systems (Blöß et al., 2017), the restriction of genetic testing (Lim et al., 2012), the attitude of doctors (Behan et al., 2016; Lim et al., 2012; Zurynski et al., 2017), the need for more psychological support (Adams et al., 2018; Anderson et al., 2013; Henderson et al., 2009), the lack of

coordination (Anderson et al., 2013; Esquivel-Sada & Nguyen, 2018; Lopes et al., 2018), research (Anderson et al., 2013; Behan et al., 2016; Christensen et al., 2017), financial support (Anderson et al., 2013; Lopes et al., 2018; Zurynski et al., 2017) or other support systems (Custódio et al., 2018; Germeni et al., 2018; Granero-Molina et al., 2020).

Finally, two relevant aspects should have been detailed. On the one hand, the obstacles for the diagnostic process and its consequences affected not only patients and families but also health professionals (Requena et al., 2014; Lopes et al., 2018). On the other hand, the health system of a country and the type of coverage it offers (through public financing or private insurance) established clear differences in health care (Esquivel-Sada & Nguyen, 2018).

4.4 | Stakeholder Consultation

Finally, 4 interviews were conducted with people affected by RD. The interviews were conducted by telephone due to the limitations of the COVID-19 situation and the territorial dispersion of the people. The main results of the interviews validate the data obtained in the review: (a) the diagnostic process as very long and distressing with presence of chronic symptomatology and sometimes very disabling to lead a normal life, along with several previous misdiagnoses; (b) they emphasised above all the emotional impact during the diagnostic process for them and their environment; (c) a significant change from the moment of diagnosis, ambivalent feelings but feeling of great psychological relief to be able to put a name to the disease; (iv) the need for patients and families to participate more actively in decision-making, as negative emotions sometimes act as a barrier; (v) the need for more support systems and they positively highlight the work of associations for those affected and their families; and finally (vi) the need for comprehensive and more humanistic care.

5 | DISCUSSION

The findings of this study detailed the emotional reactions that emerged in the diagnostic process as a response to the challenges faced by the affected people and their families plus the support elements analysed in the form of facilitators or barriers. Not having a diagnosis and the delay in diagnosis were stressors identified in all the studies in this article and in the analysis of the interviews conducted. This absence and delay generates meaninglessness, which impacts the state of mental health of both patients and their families, requiring psychosocial intervention (Kole & Faurisson, 2009).

Family caregivers felt responsible for managing the diagnostic process given the patient's health situation, leading the course of seeking the diagnosis, additional expertise and the need to make decisions. All this generates feelings of guilt and high levels of anxiety (Applebaum et al., 2020). Guilt and helplessness were a way of being anchored in the past, making it difficult to focus on the present, blocking the possibility of accepting and adapting to the situation at hand. Similarly, high levels of anxiety and uncertainty hindered

the ability to think clearly and make decisions, concerns lead to predicting a future without having enough information to support this outlook, which could endanger vital goals and values in the face of loss of control and be a threat to self-concept (Zurynski et al., 2017).

Another of the outstanding findings of this study was the feeling of confidence and security generated by having the diagnosis, apart from the uncertainty derived from the type of pathology. The diagnosis was valued as a turning point that allowed for adaptation to a new normality (Germení et al., 2018). It put an end to the process described by the presence of feelings of negative valence, whether they were high or low intensity, such as anger or deep sadness, respectively, which can lead patients and/or relatives to explode with aggressive behaviour and/or implode favouring the appearance of mood disorders such as depression (Morel & Cano, 2017). Moreover, having a diagnosis also allowed the patient to feel acknowledged and accepted, as did the support of associations and people with the same pathology, which had repercussions on their self-concept and improved their self-esteem. But in newly diagnosed people, it could be a disadvantage as it may be distressing to show the seriousness of their illness (Keary et al., 2020). Although studies such as Ryu et al., (2020) report how having the diagnosis is a motivation to change habits, they refer to it as an opportunity to become a new me.

Patients report the sudden appearance of unusual symptoms, such as feeling a difference in their body or the anxiety impact of the arrival of an unknown disease with a strange name (Ryu et al., 2020). This impact is not only personal but also social, confronting the person with a still challenging balance between equality and difference, and the search for a sense of normalcy (von der Lippe et al., 2017). This pattern of emotional responses prior to diagnosis could serve as a basis for developing psychological support tools for patients and families and for health professionals.

In the diagnostic process in general and in coherence with the results of this study, the importance of a health system and organisational factors, such as communication and care coordination or large-scale national or international initiatives to improve healthcare delivery, should be emphasised (Singh et al., 2019), as well as establishing collaboration between healthcare providers, government, politicians and society to derive more resources and more adequate management (Shafie et al., 2020).

This review in line with other studies (Depping et al., 2021) showed that it is necessary to improve health systems since participants had unmet psychological, social, personal, health and information systems, and care needs. These psychological needs require the development of individual strategies like disease management strategies and emotion regulation strategies (Depping et al., 2021).

A basic aspect that stood out was the communication between a person with an RD, his or her family and professionals, as an essential element to maintain trust during the whole diagnostic process. The need for reciprocal communication to improve opportunities for collaboration and understanding is highlighted (McMullan et al., 2020). The study by Gong et al. (2020) shows the differences in perception between doctors and patients, the former treating no more than one or two patients during their career, while the patients

instead face the challenges of their illness for life. This element of perception must be overcome.

The potential of people with an RD as experts involved with training other people, caregivers or health professionals was another element that had emerged. In the study by McMullan et al. (2020) in addressing the training of professionals, they acknowledge the potential for awareness that they can exercise and how patients can help professionals recognise the skills they already have that can be transferred to the care of a person with an RD. This patient experience also changes the traditional patient–doctor relationship, evolving into a pattern of patient-led interaction (Budych et al., 2012). The patient is an active force for change.

In line with the Granero-Molina et al. (2020) included in this review, the nurse practitioner is key to restructuring service delivery as he or she can ensure the removal of some barriers and encourage communication to provide accessible and affordable care to an RD patient (Allred et al., 2017). The nurse practitioner can play a crucial role in coordinating communication between patients, their families and all healthcare providers on the team. However, as with other healthcare professionals, it is essential to improve the level of training. Walkowiak and Domaradzki (2020) study of nursing students and professionals highlights the need for specific training and details important shortcomings. Given the genetic origin of many RDs, it is essential to pay attention to precision medicine and health care, the integration of genetic and genomic knowledge as an essential competency in nursing care, research and education (Lebet et al., 2019).

Requena et al., (2014) state that emotional stress was almost inevitable in the communication of the diagnosis. Patients narrated this moment as traumatic. The scarce knowledge of the diagnosis affected even the professionals, materialising in the use of terminology or technicalities never previously heard by the patient or family. Therefore, it is essential that professionals have guidelines and resources that can help these newly diagnosed individuals with an RD acquire appropriate coping strategies (Allred et al., 2017; Morel & Cano, 2017). As described above, professionals have a significant influence on the experiences and perceptions of patients and/or their families and can be a support or an obstacle for them (Walkowiak & Domaradzki, 2020).

Nursing plays an essential role, and they are always at the patient's side, accompanying them to improve their quality of life. RDs require clear guidelines for action, and training for nurses is necessary, as there are more than 7,000 RD characterised by a wide diversity of symptoms that can vary not only from disease to disease but also from patient to patient suffering from the same disease. Training on RD should be included in the curricula of Nursing Universities, as this would have a very positive impact on both the affected patient and their relatives (Walkowiak & Domaradzki, 2020).

The training of future professionals in the field of rare diseases is very important; nurses trained in the care process are needed to provide comprehensive care. The corresponding care must be provided in each case, taking into account the specificities of each disease and of the person him/herself at each moment. Having the opportunity to learn about cases of different RD gives us a much broader field of vision of the characteristics, course of the pathology, evolution and nursing needs in each one of them (Walkowiak & Domaradzki, 2020).

5.1 | Limitations

The main limitation is the heterogeneity of the studies in relation to the inclusion of all possible RDs. By performing a global search rather than exploring by disease, some relevant studies may have been excluded in the process. Moreover, by focusing only on the pre-diagnostic phase and the phase at the time of diagnosis, some articles may have been lost due to the difficulty of obtaining this information. In addition, the included studies were conducted in different countries and the patients' health conditions and cultural backgrounds could be different. But most of the studies were within Western culture, mainly in Europe, North America and Australia. In contrast, very little has been studied in poor countries. The reason being that in those countries, neither diagnosis nor treatment can be aspired to by most people affected by RDs. Besides, there is a lack of awareness of its importance, which is the first step in defining health, diagnostic and treatment policies (Agrawal et al., 2019).

5.2 | Implications for practice, education or research

This study revealed the need for health policy makers to contemplate the biopsychosocial impact of pre-diagnosis and timing of diagnosis, considering that individuals with an RD and their families are more vulnerable. That is why professionals need formative, informative and technological support systems.

Similarly, with the results obtained, it is suggested that a network of expert nurses should be developed to help alleviate the pressure on patients and families. The nurse can play a fundamental role as a case manager, being a link between professionals, and solving basic care needs of patients. The nurse with her/his assignment of prevention and health promotion is fundamental in the development of the autonomy of the affected persons, carrying out specific interventions that promote self-care, maintenance of daily activities, readaptation to the environment and/or emotional support.

As this article shows, the emotional consequences of the diagnostic process require emotional support for people affected by RD. This is an essential pillar of treatment. The nurse is trained to help people to manage the emotional impact to favour their adaptation and to avoid psychopathological consequences, and when it is not possible to deal with it due to the level of nursing competence, to refer them to other professionals. There is also a need to develop transdisciplinary work between health and social professionals.

Training in RD should be included in nursing education curricula, not only from the description of these processes but also from the detection of needs that can improve the quality of life of patients. Its incorporation would also help to raise visibility and awareness of these diseases through nurses. It is expected that this study will promote other research to respond to the problems identified in two aspects: one related to the development of biopsychosocial coping

strategies and the other related to improving the interventions of nursing. In addition, more future studies are needed to include more patients and diversity of RDs.

6 | CONCLUSIONS

This study provides evidence of the impact of the diagnostic process on people affected by an RD and their family members, and of the support systems in the form of facilitators and barriers. This should enable a more precise and adapted approach and provide tools for improvement of health systems and professionals in the form of information, training and resources (local, national and international). The harmonisation of global strategies should minimise the impact of socio-demographic inequalities.

Studies show the potential of affected people and their families as a key element of adaptation and normalisation. Those affected ask professionals to develop non-technical skills, used in all types of diseases, such as confidence building, communication, coordination and a comprehensive approach to professionals. In this context, the nursing professional can play a key role in coordinating communication between those affected, their families and all the healthcare and social care providers in the health team and develop diagnostic support tools. Finally, it would be important to increase research in relation to the diagnostic process to provide professionals and affected people and their families with more knowledge.

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AUTHOR CONTRIBUTIONS

Laia Llubes-Arrià: Conceptualisation, Methodology, Data review, data collection and data analyses, Writing-Reviewing and Editing, Writing-Original draft; **Montserrat Sanromà-Ortíz:** Methodology, Data analyses, Writing-Original draft, Writing-Reviewing; **Alba Torné-Ruiz:** Data review, Writing-Reviewing and Editing; **Elena Carrillo-Álvarez:** Data analyses, Writing-Reviewing and Editing; **Judith García-Expósito:** Writing-Reviewing and Editing; **Judith Roca:** Conceptualisation, Methodology, Data review and data analyses, Writing-Reviewing and Editing, Supervision.

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SUPPORTING INFORMATION

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